



SLC12A3 gene

solute carrier family 12 member 3

Normal Function

The *SLC12A3* gene provides instructions for making a protein known as NCC. This protein is a sodium chloride co-transporter, which means that it moves charged atoms (ions) of sodium (Na^+) and chlorine (Cl^-) across cell membranes.

The NCC protein is essential for normal kidney function. It is part of the mechanism by which kidneys reabsorb salt (sodium chloride or NaCl) from the urine back into the bloodstream. The retention of salt affects the body's fluid levels and helps maintain blood pressure.

Health Conditions Related to Genetic Changes

Gitelman syndrome

More than 140 mutations in the *SLC12A3* gene have been identified in people with Gitelman syndrome. Most of these mutations change single protein building blocks (amino acids) in the NCC co-transporter protein. These mutations prevent the protein from reaching the cell membrane or alter the protein's ability to transport sodium and chloride ions. Other mutations in the *SLC12A3* gene insert or delete genetic material or lead to the production of an abnormally short, nonfunctional version of the NCC protein.

Mutations in the *SLC12A3* gene impair the kidneys' ability to reabsorb salt, leading to the loss of excess salt in the urine (salt wasting). Abnormalities of salt transport also affect the reabsorption of other ions, including ions of potassium, magnesium, and calcium. The resulting imbalance of ions in the body underlies the major features of Gitelman syndrome.

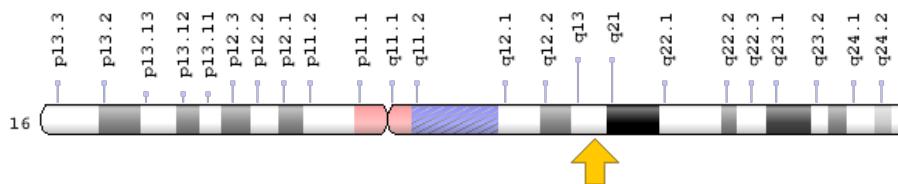
other disorders

Some research suggests that normal variants (polymorphisms) in the *SLC12A3* gene may help explain differences in blood pressure between people. Certain rare polymorphisms also appear to protect against high blood pressure (hypertension). Changes in the *SLC12A3* gene may affect blood pressure by altering the kidneys' ability to reabsorb salt into the bloodstream. However, some studies have not found any association between variants in the *SLC12A3* gene and blood pressure.

Chromosomal Location

Cytogenetic Location: 16q13, which is the long (q) arm of chromosome 16 at position 13

Molecular Location: base pairs 56,865,207 to 56,915,850 on chromosome 16 (Homo sapiens Annotation Release 108, GRCh38.p7) (NCBI)



Credit: Genome Decoration Page/NCBI

Other Names for This Gene

- Na-Cl symporter
- NaCl electroneutral thiazide-sensitive cotransporter
- NCCT
- S12A3_HUMAN
- solute carrier family 12 (sodium/chloride transporter), member 3
- solute carrier family 12 (sodium/chloride transporters), member 3
- thiazide-sensitive Na-Cl cotransporter
- thiazide-sensitive sodium-chloride cotransporter
- TSC

Additional Information & Resources

Scientific Articles on PubMed

- PubMed
<https://www.ncbi.nlm.nih.gov/pubmed?term=%28SLC12A3%5BTIAB%5D%29+AND+%28%28Genes%5BMH%5D%29+OR+%28Genetic+Phenomena%5BMH%5D%29%29+AND+english%5Bla%5D+AND+human%5Bmh%5D+AND+%22last+3600+days%22%5Bdp%5D>

OMIM

- SOLUTE CARRIER FAMILY 12 (SODIUM/CHLORIDE TRANSPORTER), MEMBER 3
<http://omim.org/entry/600968>

Research Resources

- Atlas of Genetics and Cytogenetics in Oncology and Haematology
http://atlasgeneticsoncology.org/Genes/GC_SLC12A3.html
- ClinVar
<https://www.ncbi.nlm.nih.gov/clinvar?term=SLC12A3%5Bgene%5D>
- HGNC Gene Family: Solute carriers
<http://www.genenames.org/cgi-bin/genefamilies/set/752>
- HGNC Gene Symbol Report
http://www.genenames.org/cgi-bin/gene_symbol_report?q=data/hgnc_data.php&hgnc_id=10912
- NCBI Gene
<https://www.ncbi.nlm.nih.gov/gene/6559>
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<http://www.uniprot.org/uniprot/P55017>

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